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Solid Tumour Section

Short Communication

Kidney: Renal cell carcinoma with t(X;1)(p11;p34) SFPQ/TFE3

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Huret JL. Kidney: t(X;1)(p11.2;p34) in renal cell carcinoma. Atlas Genet Cytogenet Oncol Haematol 1999;3(2)

Abstract

Review on Renal cell carcinoma with t(X;1)(p11;p34) SFPQ/TFE3, with data on clinics, and the genes involved.

Keywords
Renal cell carcinoma; chromosome X; chromosome 1; SFPQ; TFE3; MiT family

Identity

Must not be confused with the t(X;1)(p11.2;q21), also found in renal cell carcinoma.

Classification

Xp11 translocation renal cell carcinoma (RCCs) harbor gene fusions involving TFE3 transcription factor. The t(6;11) RCCs harbor a specific MALAT1 (Alpha) - TFEB gene fusion.
TFEB and TFE3 belong to the same MiT subfamily of transcription factors. Because of similarities at the clinical, morphologic, immunohistochemical, and genetic levels, the Xp11 translocation RCCs and t(6;11) RCCs are currently grouped together under the category of MiT family translocation renal cell carcinoma.

Clinics and pathology

Disease

t(X;1)(p11.2;p34) is found in a subset of Xp11 translocation RCC, median age 36 years. These tumors often have papillary architecture and clear cells with subnuclear vacuoles mimicking clear cell papillary RCC, or a nested clear cell appearance mimicking clear cell RCC.

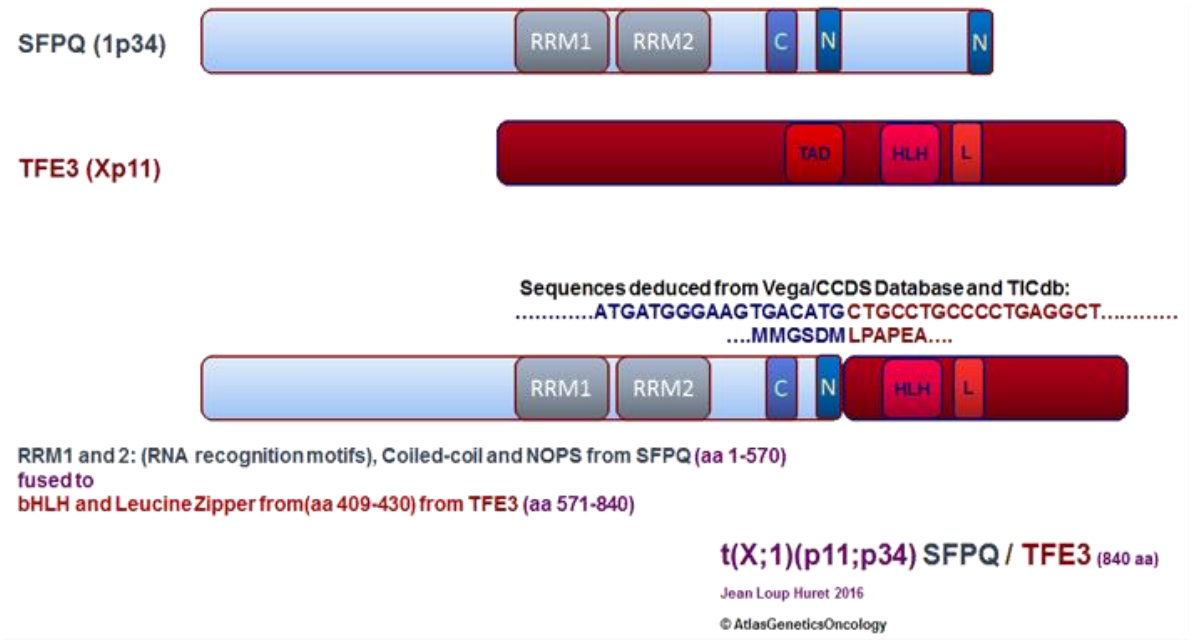
Phenotype / cell stem origin

The identical gene fusion may be found in Xp11 translocation PEComas and Melanotic Xp11 translocation cancers, which differ from the RCC in that they are PAX8 negative and Cathepsin K positive by immunohistochemistry.

Genes involved and proteins

TFE3 (transcription factor E3)

Location
Xp11.23



Protein
Contains a transcriptional activation domain , a helix-loop-helix, and a leucine zipper; member of the basic helix-loop-helix family (b-HLH) of transcription factors.

SFPQ (PTB-associated splicing factor)

Location
1p34.3

Protein
Contains RNA binding domains; involved in pre-m RNA splicing; form complexes with DNA topoisomerase I.

Result of the chromosomal anomaly

Hybrid Gene

Description
5' SFPQ - 3' TFE3

Fusion Protein

Description
N-term SFPQ and most of it fused to the DNA binding domains of TFE3 (excluding the acidic transcriptional activation domain, including the C-term helix-loop-helix, and the leucine zipper); no TFE3-SFPQ reciprocal transcript, as the der(X) t(X;1) is missing; the normal TFE3 transcript is found.

References

Argani P. MiT family translocation renal cell carcinoma. Semin Diagn Pathol. 2015 Mar;32(2):103-13

Argani P, Zhong M, Reuter VE, Fallon JT, Epstein JI, Netto GJ, Antonescu CR. TFE3-Fusion Variant Analysis Defines Specific Clinicopathologic Associations Among Xp11 Translocation Cancers. Am J Surg Pathol. 2016 Jun;40(6):723-37

Argani P, Aulmann S, Karanjawala Z, Fraser RB, Ladanyi M, Rodriguez MM. Melanotic Xp11 translocation renal cancers: a distinctive neoplasm with overlapping features of PEComa, carcinoma, and melanoma. Am J Surg Pathol. 2009 Apr;33(4):609-19

Argani P, Aulmann S, Illei PB, Netto GJ, Ro J, Cho HY, Dogan S, Ladanyi M, Martignoni G, Goldblum JR, Weiss SW. A distinctive subset of PEComas harbors TFE3 gene fusions. Am J Surg Pathol. 2010 Oct;34(10):1395-406

Dijkhuizen T, van den Berg E, Wilbrink M, Weterman M, Geurts van Kessel A, Störkel S, Folkers RP, Braam A, de Jong B. Distinct Xp11.2 breakpoints in two renal cell carcinomas exhibiting X;autosome translocations. Genes Chromosomes Cancer. 1995 Sep;14(1):43-50

Kovacs G, Szücs S, De Riese W, Baumgärtel H. Specific chromosome aberration in human renal cell carcinoma. Int J Cancer. 1987 Aug 15;40(2):171-8

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